Initiative by

PPHF
PEOPLE TO PEOPLE HEALTH FOUNDATION

Takeda

Co-powered by

Brief on North Region Rare Disease Consultation:

22 January 2021

Strategic Partners

ORD Organization for Rare Diseases India

LSDSS Lysosomal Storage Disorders Support Society

Cure SMA India

Research Partner

GRID Council

Knowledge Partner

Institute of Child Health, Calcutta

About 10% of the global population is affected by rare diseases (RDs), with 5% or less having a chance of treatment for it. Almost 80% of the rare diseases have genetic causes, affect children disproportionately, with a 30% of them dying before their fifth birthday. This mandates the need for early diagnosis, new-born screening, and awareness about these at a much larger scale. For a diverse country like India, approaches to these diseases often fall short as a result of not considering the entire ecosystem. This requires working on complex, time-sensitive actions involving multidisciplinary commitment of stakeholders to frame and implement innovative affordable solutions in an integrated manner.

After successful completion of the first virtual consultation of Eastern Region in partnership with state of West Bengal, **People to People Health Foundation (PPHF)**, and the Government of Chandigarh and Punjab jointly organized a virtual consultation on RDs for northern states of India. This consultation was the second in the series - ***"The Blueprint for Rare Disease in India initiative"***, an attempt to enhance policy commitment to prioritize access, investment, and partnerships on RDs in India. This initiative also plans to organize one more regional consultation in the Southern region. A global consultation is being organized on 28th February, the **International Rare Disease Day**. The initiative has been co-powered by Takeda Pharmaceutical and supported by the Embassy of Japan. The associate partners include ORDI, LSDSS, Cure SMA Foundation of India, RDIF (Strategic Partners), GRID Council (Research Partner) and Institute of Child Health, Kolkata (Knowledge Partner).

The objective of the consultation was to discuss and prioritise the needs of people suffering from RDs, to address challenges involved in management of rare diseases, list priority actions, explore partnership opportunities among the stakeholders and beyond for the development of better infrastructure, diagnosis, treatment, funding and comprehensive management systems. The consultation emphasised on increasing collaboration and commitment to address challenges related to RDs. The high-level recommendations that are generated from these consultations will be shared with the Government of India and Ministry of Health and Family Welfare to strengthen the upcoming ‘Rare Diseases – National Policy’.

The event opened with the welcome address by Dr. Laxmikant Palo, Chief Executive Officer, PPHF who introduced the distinguished keynote speakers of the inaugural session which included leaders from the State Governments like Dr. Amandeep Kaur Kang (Mission Director NHM, Chandigarh), Dr. K.G. Ananthakrishnan (DG, OPPI), and Dr. G.B. Singh (DoHS, Punjab). The speakers shared their insights and set the context for the consultation. During the inaugural session, an awareness video on RDs was shown to sensitize the audience through the patient’s lens. You can watch the full video by clicking on the link:
https://www.youtube.com/watch?v=zHye8nFusyQ&t=10s

This was followed by a panel discussion on *'Key unmet needs in the management of rare diseases at the State level & Priority actions for states and best practices’*. This panel discussion was chaired by - Dr. Madhulika Kabra (Professor, Division of Genetics, Dept. of Pediatrics, AIIMS). The respected panellists included Dr. Ratna Dua Puri (Chairperson, Senior

Consultant, Institute of Medical Genetics & Genomics, Sir Ganga Ram Hospital, New Delhi), Dr. Neerja Gupta (Associate Professor, Division of Genetics, Dept. of Pediatrics, AIIMS), Dr. Inusha Panigrahi (Professor, Paediatric Medicine, PGIMER, Chandigarh), Dr Seema Kapoor (Professor Paediatrics and Coordinator of Genetic Unit & Lab, MAMC & LNJP Hospital, Delhi), Dr. Ashok Gupta (Senior Professor & Medical Superintendent, Department of Paediatric Medicine, JK Lone Hospital, Rajasthan) and Dr. Shubha Rao Phadke (Professor and Head, Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow).

Post the panel discussion, other sessions were conducted. These included sessions on *the Role of Corporate sector in mobilizing better rare disease ecosystem/ Innovation in collaboration for rare diseases; PSUs- Key Stakeholder in Creating Health Avenues; Current scope of PSUs in Health Sector & potential to explore newer health areas; Journey of a parent & what it led to; Seeking multi-stakeholder support in creating a sustainable pathway for newer treatments of SMA and Rare disease policy: What is required*.

These sessions were conducted by eminent speakers like Mr. Philip Towle (Head of Access Markets Transformation, ICMEA, Takeda), Mr. Pramod K. Sinha (Formerly with Dept. of Public Enterprises, Top- Management Member of MNCS & SCOPE, New Delhi), Mr. D.P Kaushal (CGM SJVN Ltd, Shimla), Mr. Saurabh Singh (Co-Founder cum Director, RDIF), Ms. Archana Vashist Panda (Co-Founder & Director Patient Advocacy, Cure SMA Foundation of India), Mr. Manjit Singh (President, LSDSS) and Mr. Prasanna Shirol (Co-Founder & Executive Director, ORDI).

The event witnessed approximately 222participants from diverse fields including government officials of northern states, international corporations, NGOs, academia & healthcare professionals, PSUs, patient advocacy bodies etc. Full consultation recording is available at: (https://youtu.be/00-PAzNUlTE)

Key Challenges Highlighted:

- Lack of awareness about rare diseases in the community
- Inadequate knowledge and exposure among healthcare professionals including senior faculty, making it difficult to adequately train junior faculty, students, and health workers to clinical and theoretical knowledge about rare diseases.
- Limited opportunities for diagnosis at the neonatal stage and genetic testing for family members
- Challenges in availability and accessibility to treatment leading to delayed treatment and management for rare diseases where diagnosed.
- Medicines are costly and often unavailable: most of the medications are imported, due to limited domestic production
- The emotional and financial burden on patients and their families is catastrophic
- Lack of a sustainable funding mechanism through national policy for RDs
- Limited number of centres of excellence for rare diseases at the state level

High-level Recommendations:

Creating awareness

- Enrich the teaching curricula and clinical practices by including information on RDs, medical genetics and genetic counselling.
- Generate awareness among medical undergraduates, postgraduates, faculties, practitioners, PSUs, and corporates.
- Create ambassadors for RDs among medical fraternity.
- Generate more evidence on rare monogenic autosomal dominant and recessive disorders and phenotypic variants.
- Upscale therapeutic research in the niche areas e.g., gene transfer therapies, gene disruption technologies, gene modified cell therapy, and gene editing approaches.
- Build international reference network so that nations can cross-and-co-learn in a comprehensive manner and enhance implementation at the local level.
- Experience sharing by the patients, family members and patient support groups should be encouraged.

Focus on entire ecosystem

- **Intensify efforts for early identification**: Screening at prenatal, neonatal and preconception levels will help in early diagnosis and treatment. Prenatal diagnosis and preimplantation genetic testing are recommended to translate genetic and genomic advances into population health gains. Population specific issues like endogamy and consanguinity can be dealt by providing genetic counselling
- **Make services accessible and equitable**: A community-based approach for the early diagnosis, standardised treatment and risk management combined with increasing awareness is needed. This will lead to increased experience among the stakeholders including the community. At state level, patients should be recommended for definitive treatment with the approval of the medical cum technical committee of each state. The applications being considered for treatment should be blind and coded to enable an unbiased approach. Generate mechanisms to access the globally available treatment for Indian patients.
- **Evolve models of healthcare:** A Centre of Excellence for RDs based on the density of patients with specific conditions can serve as a referral and training centre nationally. Creation of a dedicated National Treatment and Care Coordination Centre would help further optimize services for RDs. It was suggested that at least one tertiary care hospital is needed in each state for diagnosis, treatment and management of RDs. Hub and spoke models can be developed with a minimum 4-5 spoke hospitals in a state for regular care.
- **Amplify awareness among general public:** Commemorate ‘World Rare Disease Day” every year, and give it the same weightage in the public discourse as other such days.

"Rare disease- National Policy" once implemented must be reviewed periodically along with the list of conditions included in the current national registry for RDs.

Success stories:

- The motivating journey of the *Rare Disease Centre of Excellence* and the *Public Education Program* run by the J K Lon hospital, Jaipur, **Rajasthan** was shared in great details. Initiated in 2014, the patient reporting with cases like metabolic, dystrophic and distal muscular dystrophy (DD) have significantly increased by the year 2019. To further increase the awareness about the issue under the public education program, a book named '*Rare Diseases in Pediatrics*' was published in collaboration with the Indian academy of pediatrics (IAP), a course manual on rare disease was designed to sensitize pediatricians across India. A multi disciplinary team consisting of physicians and state government officials with similar interests was established as 'working groups' to further widen the sensitization about the issue. The centre of excellence, Jaipur is equipped with a 12-bedded rare disease ward with infusion facility and a new-born screening lab which has successfully screened approximately 30,000 new-borns in 2019 itself, by the support of the state government and CSRs. The rare disease web registry India was also framed. As a part of this program, the centre was represented at various international meetings including the *First International Rare Disease Day, Primary Immunodeficiency Conference 2018, Faculty World Symposium Rare Diseases*. The centre collaborated with various medical colleges in the state including AIIMS Jaipur to work on projects like DBT NIDAN Kendra launch, Genome India project. To direct the social, political and scientific awareness, a logo with human formation was also released. These activities together helped in early disease detection such as SMA with pompe, early management measures such as Risdiplam for SMA and giving a base for future studies as well.

- Success stories from the state of **Uttar Pradesh** had also given a direction to the discussion. To address the need for training the doctors, the state has adopted a 2 weeklong program supported by the ICMR that has successfully trained more than 100 clinicians under approximately 18 programs in the last 20 years. Once the patients are detected, treatment availability is often assured in terms of adequate logistics and cost, by the *Patient support groups.* Examples quoted were Hemophilia Federation of India, Thalassaemic India etc who made possible the availability of free treatment for the patients of hemophilia and thalassemia by working/motivating policy makers under the government supported programs.

- The role of public sector units (PSUs) was magnificently specified by the SJVN, Solan, **Himachal Pradesh** which has been constantly working with the Indian association of muscular dystrophy, Solan and has financially aided in the establishment of the *Muscular Dystrophy Centre.* The centre has organized 114 medical camps for nearly 1785 patients and benefitted approximately 884 patients since 2017.

The Indian Rare Disease Registry, an initiative by ICMR ought to be considered as a success as it rolled out in the phase manner. The first phase included Haemoglobinopathies, treatable Lysosomal Storage Diseases, small molecule Inborn Errors of Metabolism, Coagulopathies, Neuromuscular Disease like Duchenne

- Muscular Dystrophy, Spinal Muscular Atrophy, common Primary Immunodeficiencies, common Skeletal Dysplasias to name a few.

- To address the catastrophic expenditure on the families of patients with rare diseases, the government has proposed a financial support of up to ₹15 lacs under the umbrella scheme of the Rashtriya Arogya Nidhi for those who require one-time treatment. The beneficiaries for such financial assistance extends from below poverty line(BPL) to 40% of the population eligible as per the norms of the Pradhan Mantri Jan Arogya Yojana, for the treatment in government tertiary hospitals only.

For more Information contact:

Ms. Cheena Malhotra: cmalhotra@phfindia.org Phone no.: 9650516668
Ms. Megha Gupta: mgupta@phfindia.org Phone no.: 9311718579